Note: The following information is provided by the author(s) and has not been reviewed by *GeneReviews* staff.

Table 2. Selected *MCOLN1* Mutations Not Discussed in Mucolipidosis IV *GeneReview*

c.163_197del, c.163_197insTCA
c.473_474delCC
c.1209-1210insT
c.1463_1464insGGCCGCAGCAG [2]
c.304C→T
c.514C→T
c.964C→T
c.317T→C
c.497G→T
c.1207C→T
c.1336G→T
c.1340T→C
c.1395C→G
c.1388G→A
c.236_237ins93 from NADH dehydrogenase 5 99-192 [1]
c.302_303delTC
c.920delT
c.1614delG
g.12426 A→T
c.235C→T

1. An American and a Canadian family was found to have a 93-bp insertion mutation in exon 2 that leads to altered splicing and null expression. The inserted segment was identical in sequence to part of the gene encoding mitochondrial NADH dehydrogenase subunit 5 [Goldin et al 2004].
2. An 11-nucleotide insertion mutation causing a frame shift in exon 10 was discovered in two non-Jewish individuals [Bargal et al 2001, Altarescu et al 2002].

Altarescu G, Sun M, Moore DF, Smith JA, Wiggs EA, Solomon BI, Patronas NJ, Frei KP, Gupta S, Kaneski CR, Quarrell OW, Slaugenhaupt SA, Goldin E, Schiffmann R (2002) The neurogenetics of mucolipidosis type IV. *Neurology* 59:306-13

Bargal R, Avidan N, Olender T, Ben Asher E, Zeigler M, Raas-Rothschild A, Frumkin A, Ben-Yoseph O, Friedlender Y, Lancet D, Bach G (2001) Mucolipidosis type IV: novel MCOLN1 mutations in Jewish and non-Jewish patients and the frequency of the disease in the Ashkenazi Jewish population. *Hum Mutat* 17:397-402

Goldin E, Stahl S, Cooney AM, Kaneski CR, Gupta S, Brady RO, Ellis JR, Schiffmann R (2004) Transfer of a mitochondrial DNA fragment to MCOLN1 causes an inherited case of mucolipidosis IV. *Hum Mutat* 24:460-5